

## CLINICAL AND RADIOLOGICAL FINDINGS IN ARNOLD CHIARI MALFORMATION

Abdul Aziz Khan, Sajid Nazir Bhatti, Ghayyur Khan\*, Ehtisham Ahmed, Ahsan Aurangzeb, Asghar Ali, Amjad Khan, Saleem Afzal

Department of Neurosurgery, \*Radiology, Ayub Medical College, Abbottabad, Pakistan

**Background:** The Chiari Malformation I (CMI) is a disorder of uncertain origin that has been traditionally defined as downward herniation of the cerebellar tonsils through the foramen magnum. The anomaly is a leading cause of syringomyelia and occurs in association with osseous abnormalities at the craniovertebral junction. In contrast to other Chiari malformations, CMI tends to present in the second or third decade of life and is sometimes referred to as the 'adult-type' Chiari malformation. The objective was to document clinical and radiological findings in Arnold Chiari Malformation-I. **Method:** This was a descriptive study carried out in Ayub Teaching Hospital Abbottabad at Neurosurgery Department during July 2008–July 2010. We examined a prospective cohort of 60 symptomatic patients. All patients underwent magnetic resonance imaging of the head and spine. **Results:** There were 40 female and 20 male patients. The age of onset was  $24.9 \pm 15.8$  years. Common associated radiological problems included syringomyelia (60%), scoliosis (25%), and basilar invagination (12%), increased cervical lordosis 5 (8.5%), and Klippel Feil syndrome 2 (3.3%). The most consistent magnetic resonance imaging findings were obliteration of the retrocerebellar cerebrospinal fluid spaces (70% patients), tonsillar herniation of at least 5 mm (100% patients), and varying degrees of post fossa anomalies. Clinical manifestations were headaches, pseudotumor-like episodes, a Meniere's disease-like syndrome, lower cranial nerve signs, and spinal cord disturbances in the absence of syringomyelia. **Conclusion:** These data support accumulating evidence that CMI is a disorder of the para-axial mesoderm that is characterised by underdevelopment of the posterior cranial fossa and overcrowding of the normally developed hindbrain. Tonsillar herniation of less than 5 mm does not exclude the diagnosis. Clinical manifestations of CMI seem to be related to cerebrospinal fluid disturbances (which are responsible for headaches, pseudotumor-like episodes, endolymphatic hydrops, syringomyelia, and hydrocephalus) and direct compression of nervous tissue.

**Keywords:** Arnold Chiari Malformation, Posterior cranial fossa, Cerebrospinal fluid

### INTRODUCTION

The Chiari Malformation I (CMI) is a disorder of uncertain origin that has been traditionally defined as downward herniation of the cerebellar tonsils through the foramen magnum.<sup>1</sup> The anomaly is a leading cause of syringomyelia<sup>2</sup> and occurs in association with osseous abnormalities at the craniovertebral junction. In contrast to other Chiari malformations, CMI tends to present in the second or third decade of life and is sometimes referred to as the 'adult-type' Chiari malformation.

Chiari II malformation, which is present at birth and consists of downward herniation of the lower cerebellum and medulla into the spinal canal, in association with myelodysplasia and complex anomalies of the brain such as aqueductal forking and polymicrogyria.<sup>3</sup> The comparatively rare Chiari III malformation is also present at birth and is defined as cerebellar herniation into a cervical encephalocele.<sup>4</sup> From a developmental standpoint, Chiari malformations have been classified as neuroectodermal defects<sup>3</sup>, although there is accumulating evidence that tonsillar herniation in CMI is attributable to overcrowding of the hindbrain by an underdeveloped posterior cranial fossa (PCF).<sup>5</sup> Current interest in CMI can be attributed to magnetic resonance imaging (MRI), which has

revolutionised diagnosis and has led to the detection of cases that either were not recognised or were erroneously identified as other conditions. The resulting increase in the number of reported cases has emphasised the need for greater understanding of the pathogenesis and clinical manifestations of CMI. Growing concerns regarding the risk of inheritance cannot be answered on the basis of case reports of familial aggregation.<sup>6</sup>

To investigate the syndrome of CMI, we studied a prospective cohort of 100 symptomatic patients, including those with ambiguous or unfamiliar symptoms. The study provides a comprehensive review of the symptoms, neurodiagnostic findings, in this group of patients.

### MATERIAL AND METHODS

The study was carried out in Ayub Teaching Hospital Abbottabad at Neurosurgery Department during July 2008–July 2010, patients presented to Outpatient Department were included in the study. Out of 60 eligible patients, 40 were female and 20 were male, each patient was symptomatic and had MRI findings consistent with CMI or an anomaly of the basicranium in association with syringomyelia, brain and osseous structures were assessed qualitatively.

The slope of the tentorium and the length of osseous structures were defined subjectively as normal, increased, or decreased, based on measurement parameters for 50 control subjects. We defined odontoid retroflexion as in the almost circular space bounded by the tentorium, occipital bone, clivus, and petrous bone.



**Figure-1: Tonsillar herniation less than 5 mm**



**Figure-2: Tonsillar herniation more than 5 mm**

## RESULTS

Ocular disturbances were reported by 45 patients 75%. Affected individuals were defined as those with two or more of the following intermittent symptoms: retro-orbital pressure or pain, visual phenomena such as floaters or flashing lights, blurred vision, photophobia, diplopia, and visual field cuts. Most of these symptoms were accentuated by the same factors that affected suboccipital headaches. Neuro-ophthalmological examinations revealed few objective findings. Ocular disturbances among young women were the leading cause of erroneous diagnoses of multiple sclerosis.

A total of 20 patients (33%) experienced otoneurological disturbances that included two or more of the symptoms: dizziness, disequilibrium, pressure in

the ears, tinnitus, decreased hearing or hyperacusis, vertigo, and oscillopsia. Like ocular disturbances, most of these symptoms were accentuated by the same factors that affected suboccipital headaches. There were very few objective findings except for nystagmus. Complete otological examinations were performed for 8 patients with disabling dizziness or vertigo, i.e., a peripheral type characterised by impaired caloric responses and the absence of central abnormalities (12 patients) and a central type characterised by normal caloric responses and distinct neural abnormalities, such as impaired optokinetic nystagmus, impaired smooth pursuit, saccadic dysmetria, and downbeat, positional, or periodic alternating nystagmus (2 patients). Clinical examination reveals lower cranial nerve, brain stem, and cerebellar disturbances for 40 (66.6%). The most common symptoms were dysphagia, sleep apnoea, dysarthria, tremors, palpitations, and poor coordination. Objective findings consisted of cranial nerve deficits and cerebellar signs in a majority of the patients with the Chiari I Malformation.

Disturbances of spinal cord function were present in 50 patients (84%) with syringomyelia. Most common symptoms were muscular weakness, paresthesia or hyperesthesia, nonradicular segmental pain, analgesia or anaesthesia, spasticity, trophic phenomena, burning dysesthesia, and poor position sense. Objective findings included impaired fine-motor function of the hands (48 patients), muscular weakness (25 patients), analgesia or anaesthesia (18 patients), and hyperreflexia (18 patients).

The most constant findings were compression of the CSF spaces posterior and lateral to the cerebellum tonsillar herniation of at least 5 mm, reduced height of the supraocciput (30, 50%), and increased slope of the tentorium (10, 16.6%). Other findings included reduced length of the clivus, anterior displacement of the cerebellum, kinking of the medulla, retroflexion of the odontoid process, basilar invagination, compression of the 4<sup>th</sup> ventricle, empty sella, hydrocephalus, and syringobulbia. Minimal evidence of hindbrain overcrowding consisted of obliteration of the retrocerebellar CSF spaces in association with a meniscus sign at the lower pole of the cerebellar tonsils. Spinal abnormalities associated with CMI included syringomyelia (35, 58.3%), scoliosis or kyphosis (15, 25%), increased cervical lordosis (5, 8.3%), and Klippel-Feil syndrome (2, 3.3%).

## DISCUSSION

Chiari malformations are generally regarded as a pathological continuum of increasingly severe hindbrain maldevelopments.<sup>7</sup> The association of Chiari II and Chiari III malformations with embryological defects of the brain and spinal cord has established these lesions as primary neural anomalies.<sup>8</sup> However, there is clinical

and experimental evidence that chronic tonsillar herniation in CMI could be attributable to underdevelopment of the occipital bone and overcrowding of the cerebellum within a too-small PCF.<sup>9</sup> Recent morphometric studies are consistent with this view, and Nishikawa *et al*<sup>10</sup>, suggested that the fundamental defect may involve underdevelopment of the occipital somites originating from the para-axial mesoderm.

Other MRI findings provided substantial evidence of hindbrain overcrowding. The most constant abnormality was compression of the CSF spaces posterior and lateral to the cerebellum (50 patients, 83.5%). Tonsillar herniation of at least 5 mm below the foramen magnum was present in 60 patients (100%). Less constant findings of hindbrain overcrowding included anterior displacement of the cerebellum, kinking of the medulla occurring with retroflexion of the odontoid process or basilar invagination, and compression of the fourth ventricle. There is limited information regarding the epidemiological features of CMI. The anomaly is defined as a rare disorder<sup>11</sup>, and female subjects outnumber male subjects by a wide margin. approximately 25% of patients cited trauma as the precipitating factor. The most common mechanisms were whiplash injuries and direct blows to the head and neck, which raises the possibility that certain types of trauma accentuate tonsillar impaction or result in subarachnoid hemorrhage that destabilizes a marginally compensated CSF system. The likelihood that CMI can be genetically transmitted has been suggested by two lines of evidence, i.e., the association of CMI with known genetic disorders, such as achondroplasia, Hadju-Cheney syndrome, and Klippel-Feil syndrome, and case reports of familial aggregation, including cases of monozygotic twins and triplets concordant for CMI. The incidence of familial syringomyelia is reported as 2%, but there are no data, to our knowledge, regarding the risk of inheritance in CMI. These modes of transmission have also been reported for patients with familial syringomyelia.<sup>12</sup>

Previous reports have documented the complex symptom patterns for CMI.<sup>13</sup> Presenting symptoms can include headaches<sup>14</sup> in association with a wide variety of ocular<sup>15</sup>, otoneurological, brain stem, and spinal Cord<sup>16</sup> disturbances. Although many of the symptoms discussed in this report have been described previously, some are less familiar or have been difficult to relate to the specific effects of tonsillar herniation and syringomyelia. A majority of the patients in the study population complained that their symptoms had been ascribed to psychogenic causes.

There is evidence that some ambiguous or unfamiliar symptoms may be CSF-related. For example, given the significant reduction of CSF volume in the PCF demonstrated in this study (mean: 10.8 ml, 40%), it

is evident that newly formed CSF is displaced from the compressed subarachnoid spaces of the PCF into available spaces within the supratentorial and spinal compartments. Such displacements almost certainly affect CSF compliance and would be expected to alter the normal damping effect of an open CSF system, which mitigates changes in venous volume and pressure occurring with respirations, the cardiac pulse, Valsalva manoeuvres, and changes in posture. Under these conditions, CSF displacements could play a role in the following symptoms: sub-occipital headaches that radiate to the vertex and behind the eyes and inferiorly to the neck and shoulders, pseudotumor-like episodes of retro-orbital pain and visual phenomena, and a Meniere's disease-like syndrome of pressure in the ears, dizziness, disequilibrium, tinnitus, and hearing loss. The exquisite sensitivity of these symptoms to physical activities, including Valsalva manoeuvres, is consistent with reduced compliance of the CSF system. For some patients with vestibular dysfunction, otological testing revealed low-frequency sensorineural hearing loss in association with peripheral vestibulopathy. These findings fulfil the criteria for endolymphatic hydrops and suggest that CSF displacements in CMI contribute to disturbances of CSF-perilymph dynamics.

The most obvious CSF-related symptoms are those attributable to syringomyelia. These can occur because of stretching and distension of nervous tissue or dissection of central canal cavities into the parenchymal tissues of the spinal cord. Although it was once thought that syringomyelia was caused by the forceful diversion of CSF from the fourth ventricle into the central canal, it is now known that most syrinxes do not communicate with the fourth ventricle and are separated from it by an occluded or stenotic segment of the canal.

Current evidence suggests that syrinx formation is the result of an obstruction of CSF flow at the foramen magnum, which exaggerates the pulsatile systolic pulse wave in the spinal subarachnoid space and drives CSF through anatomically continuous perivascular and interstitial spaces into the central canal of the spinal cord. More severe obstructions of CSF circulation are occasionally causes of hydrocephalus.

An unexpected finding in this study was the presence of spinal cord disturbances in 21 out of 60 patients (35%) who did not have syringomyelia. Some of these disturbances are related to mechanical compression of the cervicomedullary junction, but another explanation could be that the exaggerated systolic pulse wave in the spinal canal is capable of producing symptoms that mimic those of syringomyelia. Compression of the brain stem and lower cranial nerves was the most likely cause of dysphagia, hoarseness, sleep apnoea, and palpitations. The incidence of cardiac irregularities in the study population seemed unusually high for a cohort of predominantly young adult female

subjects, and an association between sinus arrhythmia and CMI was noted previously. It is acknowledged that many of the symptoms described in this report occur with variable frequencies in the general population and could be unrelated. Clinical descriptions of CMI have undergone continuous revision since the original report, by Chiari<sup>17</sup>, of tonsillar herniation in patients dying as a result of hydrocephalus. In recent years, the term CMI has been used synonymously with tonsillar ectopia or chronic tonsillar herniation in a wide variety of congenital and acquired disorders. The radiological definition of CMI has been reported as tonsillar herniation of at least 3 mm<sup>18</sup> or at least 5 mm<sup>1</sup> below the foramen magnum. However, this definition is limited to a single criterion and makes no reference to clinical symptoms or the presence or absence of associated findings such as syringomyelia. The radiological definition of CMI may be too restrictive. In our study almost 100% of patients have tonsillar herniation more than 5 mm. All patients showed MRI evidence of hindbrain overcrowding, and CINE-MRI demonstrated abnormalities of CSF velocity/flow that were similar to those reported for patients with tonsillar herniation of at least 5 mm.<sup>19</sup> These observations indicate that the extent of tonsillar herniation cannot be used as the sole criterion for the diagnosis of CMI. We could not confirm reports that the severity of symptoms is directly related to the extent of tonsillar herniation. Because tonsillar herniation of at least 5 mm can be encountered as an incidental finding among asymptomatic patients, it is likely that the position of the cerebellar tonsils, although providing a general index of hindbrain overcrowding, is only one factor influencing the clinical features of CMI.

## CONCLUSIONS

CMI is a disorder of the mesoderm and is thus inherently different from Chiari II and Chiari III malformations. The anomaly occurs sporadically but can be transmitted genetically in some families. The most constant feature of CMI is a volumetrically small PCF, which predisposes patients to hindbrain overcrowding. Displacements of CSF probably contribute to the symptoms.

The clinical syndrome of CMI is characterised by headaches, pseudotumor-like episodes, a Meniere's disease-like syndrome, lower cranial nerve signs, and spinal cord disturbances even in the absence of syringomyelia. Diagnosis is established by MRI. Minimal evidence of hindbrain overcrowding consists of obliteration of the retrocerebellar CSF spaces in

association with a meniscus sign at the lower pole of the cerebellar tonsils. CINE-MRI can be helpful in demonstrating a disturbance of CSF velocity/flow at the foramen magnum in patients with tonsillar herniation of less than 5 mm.

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## Address for Correspondence:

**Dr. Abdul Aziz Khan**, Department of Neurosurgery, Ayub, Teaching Hospital Abbottabad, Pakistan. **Cell:** +92-300-9176146.